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Prader Willi Syndrome – 'A Family's Experience'

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Abstract

Prader Willi Syndrome – 'A Family's Experience'

Genetic research has offered, and continues to offer, a medical explanation of chromosomal disorders such as Down Syndrome and Asberger Syndrome and more recently the rare chromosomal disorder Prader Willi Syndrome. This research gives a pathogenic explanation of disorders which includes historical background, genetic defects and clinical features. This study set out to offer an insight into the effects of PWS on the child and his/her family. It also aimed to highlight what support systems are in place in the Co. Louth area of Ireland for individuals with PWS. Unfortunately, PWS is not curable at this time. Between 1995 and 2003 there were 39 diagnosed cases of PWS in Ireland, an average of 4.3 per year. On average there are four infants or children diagnosed in Ireland with PWS per year. (Turner, 2004, National Centre for Medical Genetics).

This study is an exploratory and descriptive case study. This case study drew on multiple sources of evidence to construct a valid and unique illustration of PWS. The primary source of data was derived from in-depth interviews with the parents of a 3-year-old girl who has PWS. She was diagnosed during the third week of life. The evidence of this study suggests that non-specialist medical staff are *not* generally familiar with PWS. Training in relation to diagnostic criteria for chromosomal disorders would be extremely beneficial to them and to families that are affected by the syndrome. This study highlights the need for parents to be their own child's advocate in obtaining desired support services in their area. Support Services in the North East region have been greatly increased due to the setting up of the North Eastern Health Board (now known as Health Service Executive, North East Region) Early Intervention Services (EIS), in 2000.

Introduction

There has been a very gradual shift in Irish society's understanding of disability. This shift has involved the move away from the institutional model of care to a more holistic community based one. The type of disability this work is concerned with is both physical and intellectual. The Disability Database (2002) report that there is a prevalence rate of 7.38 per 1,000 of the total population effected by intellectual disability, in Ireland. The National Association for People with Intellectual Disabilities in Ireland NIAMHI (2001) identify five main causes of intellectual disability. They include prenatal, perinatal and postnatal factors, illness or injury to the mother during pregnancy, and lastly genetic factors. This piece of work is concerned with the genetic cause of intellectual disability.

Like Down's Syndrome, which is probably the most well known genetic syndrome, PWS is also caused by chromosomal abnormality - a deletion of part of chromosome 15.

PWS is defined as '*A chromosomal disorder resulting in a syndrome characterised by infantile hypotonia, hypogonadism and obesity.*'¹ There is a decrease in hypothalamus function of the brain which results in disturbances in appetite, satiety and the release of growth hormone. This in turn leads to an insatiable appetite for food and growth deficiency. Individuals with PWS require '*typically only 40 - 70% of calorie intake required by non-obese otherwise healthy individuals*' (Hauffa, 2000: 5).

¹ Hypotonia weak muscle tone, and floppiness at birth. Hypotonia is an early and persistent feature of the syndrome. It mainly affects the neck in the neonatal phase.
Hypogonadism immature development of sexual organs and/or other sexual characteristics.
Obesity a person can be described as morbidly obese with a body mass index (BMI) of 42 - 47 kg/m² as compared to a normal BMI of 21 - 27 kg/m². In some cases this would mean a person is double their optimal weight for their height. (www.pwsa-uk.com/en/00uk/02z.htm), 2000, PWSA UK, A-Z of Prader-Willi Syndrome).

The epidemiological facts of PWS include an incidence of 1 in every 15,000 of live births, compared to that of 1 in every 600 for Down Syndrome. At present there is no cure or pre-natal diagnosis. On average 3– 4 children are diagnosed per year with PWS in Ireland. The Prader Willi Syndrome Association of Ireland, (PWSA, Ireland) have been in contact with 64 families affected by the syndrome since their establishment in 1977. The oldest known person with PWS in Ireland is 39 years old (source: PWSA Ireland). The oldest known living person with PWS globally is a 71 year old man in the United States.

Physical characteristics of PWS which are variable, include very small hands and feet, almond shaped eyes, down turned mouth, short stature (due to growth hormone deficiency), hypo pigmentation – *'where by children show fairer skin, hair and eye colour than their parents'* (Hauffa, 2000: 4).

Secondary disorders in PWS are common. Sleeping disorders such as sleep apnoea – (cessation of breathing during sleep), which is known to be more prevalent in obese individuals with PWS, respiratory problems (due to hypothalamic dysfunction), diabetes mellitus – *'The usual cause of diabetes in PWS is obesity'* (PWSA, UK).

Behavioural characteristics in PWS vary greatly in the early and later stages. In the early neonatal phase of PWS the infant is described as sleepy and passive. This is due to the infant being hypotonic (weak muscle tone). The childhood phase (between 2 and 3 years of age) is characterised by the onset of uncontrolled hyperphagia (insatiable appetite for food). Children can begin to exhibit behavioural problems, with outbursts of temper in response to frustration of constantly craving food.

Support services for parents of a child with PWS are fundamental in managing the syndrome. Hauffa (2000: 11) cites diet as the cornerstone of management in

PWS. This is due to many of the behavioural difficulties associated with PWS stemming from the constant drive for food.

The multi faceted approach which involves several relevant health professionals working together on particular cases has slowly become embedded in Early Intervention Services in Ireland. Formal establishment of such services begun in 2000 and 2001 and is still ongoing. At present there is no specific policy or legislation that focuses solely on Early Intervention Services. However it is posited in the wider context of disability legislation. The Strategy of Equality (Commission on the Status of People with Disabilities, 1996) highlighted the need for partnership and consultation with people with disabilities. The Government Health Strategy – Quality and Fairness, 2001, introduced the principle of people centred services and sought the development of standards in disability services. The aim of early intervention in the context of disability is to minimise the impairment effect of disability, ensure people reach their potential and support families. The Early Intervention Team in Co. Louth was formally established in 2001. This team currently work with two children with PWS, aged three and four.

Diagnostic criteria for PWS were published in 1993 in the University Of Washington School Of Medicine, Seattle. Geneticists and allied medical professionals in Ireland, United Kingdom, Canada, United States and other countries have adopted these criteria. In brief it comprises of major criteria such as neonatal and infantile hypotonia and minor criteria such as characteristic behavioural problems – temper tantrums and tendency to be oppositional. A scoring system is used to weigh up various criteria to decide on diagnosis. (Holm, V.A. *et al*, 1993)

As stated above, PWS is relatively rare in comparison with other genetic disorders such as Down's syndrome. However, many of the behavioural and social

characteristics of the syndrome are similar to that of other disorders that social care workers come in contact with, such as Autism and Asperger's syndrome.

At present there is one residential unit in Ireland which specifically cares for adults with PWS. It is situated in Leopardstown, Dublin and is funded by the Eastern Regional Health Authority. It is staffed by a Unit Leader, Team Leader and six care assistants, all of which have received specialised training from the UK Prader Willi Syndrome Association. Since its opening in 2003, three people with PWS have taken up residence there. Other adults with PWS in Ireland reside in the family home or in state/voluntary run group homes, whereby other residents have a range of learning disabilities. There is continuous debate in the field of PWS whether adults with PWS have a better chance of reaching their potential in specialist PWS care or mixed care.

Methodology

Multiple sources of evidence were used in this study. Semi structured and in-depth interviews were carried out. Due to some health professionals involved in the study not being available for interview, correspondence ensued via the mediums of email, telephone and post.

Participants

The subject of this case study is a three year old girl. In the interest of confidentiality a pseudonym, Anne, will be used to refer to her throughout this paper. Both her mother and father were the two key participants in this case study. They will be simply referred to as F, for father and M, for mother.

Other participants include the Information Officer from Prader Willi Syndrome Association Ireland, Paediatrician, National Children's Hospital, Tallaght, Genetics Counsellor, National Genetic Centre, Our Lady's Hospital for Sick Children, Crumlin, and the Coordinator of Early Intervention Services, North Eastern Health Board.

Materials

The first interview was a semi structured in-depth one with Anne's mother and father. A twenty-five-item schedule was used; items such as time of diagnosis, support services and range of effects of the syndrome were explored. The second interview was a semi-structured interview with the Information Officer from Prader Willi Syndrome Association Ireland. This interview had four fixed items, formation of support group, referral procedure, statistics of group and services. The third interview was carried out with a Paediatrician, National Children's Hospital, Tallaght. A seven-item schedule was used covering three areas, growth hormone treatment, current sleep studies with PWS, and current and oncoming treatment for individuals with PWS.

Secondary data was sourced from a Genetics Counsellor, National Genetic Centre, Our Lady's Hospital for Sick Children, Crumlin. A five item questionnaire was used covering two areas, genetic counselling and epidemiology of PWS. Secondary data was also sourced from Coordinator of Early Intervention Services, North Eastern Health Board HSE, North East Region. A nine-item questionnaire was used covering three areas, formation of Service, statistics of PWS service users, and services provided.

Procedure

Initial permission was sought for interviewing and audio recording by telephone for the first and second interviews. This was followed by letters of confirmation in November 2003. A phone interview was carried out with the Respiratory Paediatrician, National Children's Hospital, on 3 February 2004, due to her unavailability thereafter. Information was sought from the Genetics Counsellor, National Genetic Centre and from the Coordinator of Early Intervention Services via telephone, email and in writing. A Sanyo Talk book VAS was used to audio record the first and second interviews.

Ethics

A key ethical consideration of this study was confidentiality of the family concerned. The family were consulted prior to the interviewing procedure about how the researcher would ensure confidentiality. The parents were debriefed after interview. They were informed that all transcripts, information given by them will be kept securely until research has been completed and that it will be discarded thereafter. Another ethical concern was the possibility of sensitive issues being raised in interview with the family. An effort was made to word questions in an objective and open fashion, e.g. when attempting to gain an insight into whether the Anne had challenging behaviour in relation to food, parents were asked '*Does Anne have any particular difficulties in relation to wanting food?*'

Results and Discussion

What is presented here in each section is brief results and discussion on some of the key findings of this case study. The themes selected for discussion were prominent issues that presented during the core interview with Anne's mother and father.

Diagnosis

With the inception of diagnostic criteria for PWS in 1993 the syndrome is increasingly being detected and diagnosed in infancy and childhood. Anne was diagnosed during her third week of life in Our Lady's Hospital for Sick Children in Crumlin, Dublin.

The focus of the theme of diagnosis here is the effect of this time on the parents. Both parents were asked simultaneously of their experiences at the time of Anne's diagnosis.

Anne's mother, M, stated

'The nurses were so nice; we were taught how to use a special bottle to feed Anne, a Habermann bottle'. Anne's father F commented - 'from a medical point of view, the support was fine, had we been suicidal, not that we were, but...M - 'Yes, we've always thought that.... for example the way we were told about the diagnosis.... Dr. ____ brought us down to his room, there was a nurse sitting behind us, he put the two of us down on a seat. He gave us the diagnosis and then handed us a load of stuff from the Internet. F - He didn't know anything about it' (Prader Willi Syndrome).

Hauffa validates Anne's parents comments by stressing that 'all members of the immediate family require support and counselling' (Hauffa, 2000: 12). The key finding of this theme is that medical staff may not have previous knowledge of PWS prior to a case like Anne's. This will inevitably effect how they deliver information and support parents at this time.²

Genetic Counselling

Genetic counselling is generally provided for families when developmental and/or physical problems such as Down's Syndrome and PWS are detected following the birth of a child. It is also used when a diagnosis of a hereditary disease such as cystic fibrosis or Huntington's disease is made. Turner, Genetics Counsellor, 2003 states: *'genetic counselling is now a standard practice, for parents, who have recently received a diagnosis of any genetic disorder. We are happy to see families if they wish and for their children to be seen again by a clinical geneticist.'*³ The American Society of Human Genetics (1975) defines genetic counselling as:

*'An educational counselling process for individuals and families who have a genetic disease or who are at risk for such a disease. Genetic counselling is designed to provide patients and their families with information about their condition and help them make informed decisions.'*⁴

² Hauffa, Dr B., (2000) Prader Willi Syndrome, A Clinical Overview, Colwood Healthworld UK.

³ Turner, J, Genetics Counsellor, National Genetic Centre, Crumlin, email correspondence, 8 November 2003.

⁴ <http://www.medicinenet.com/script/main/art.asp>- American Society of Human Genetics, Definitions, 2001 [Accessed 16 February 2004].

Feelings of guilt can often be associated with parents of children with genetic conditions like PWS. Questions like, *'Why our family? What did I do to cause this? Will it happen again?'* can arise for parents. Anne's mother stated early in the interview *'I just didn't want to believe this could be genetic'.*

Latson (1995) writes: *'Internal stress factors come from within the individual...the first thought is What did I do wrong? Therefore, parents must learn how to develop realistic expectations and how to recognize negative self-talk.'*⁵

Anne's parents did not receive genetic counselling. They were scheduled to meet with a genetics counsellor in Crumlin hospital shortly after diagnosis but were not contacted regarding this. Anne's mother seemed to have a very clear recollection of this time. She expressed the relief she had felt when their local Health Board services NEHB (now HSE North East Region) were put in place at this time. This included early intervention services, physiotherapy and speech therapy. The key finding of this theme of genetic counselling is that although this service is now deemed standard practice after diagnosis in Ireland, it may not always be provided. This emphasises that the parents of a child with PWS need to be strong advocates for their child in relation to accessing services they are entitled too.

Early Intervention Support Services

The key concept of early intervention is to intervene early enough to make programmes implemented, into a way of life for the child. There is preventative work as well as therapeutic work carried out. Ann Hynes, Co-ordinator of Early

⁵ Latson, S.R., (1995) Preventing Parent Burn Out: Model for Teaching Effective Coping Strategies to Parents of Children with Learning Disabilities, Learning Disabilities Association of America, Online. Available from: http://www.idonline.org/id_indepth/parenting/preventburnout.htm [Accessed 11 Jan 2004]

Intervention Services in the HSE North East Region explained the make up of their EIS team and the process involved in working with children with disabilities:

'The team consists of myself, the Coordinator, two physiotherapists, a social worker, a speech and language therapist, an educational psychologist, and a nurse/counsellor. At present we are awaiting the appointment of an occupational therapist. After assessment the child will receive the services that have been identified based on our resources. We will also refer children to other services such as audiology, dental, vision within the North Eastern Health Board (now known as the HSE North East Region) and may also refer to other services outside the NEHB such as the feeding clinic in Temple Street Hospital.'

Anne is one of two children with PWS that the North East Service is currently working with. Since the early intervention service is made up of a team of people, both parents in this case study were asked about who they contact etc with queries. Both parents responded.

M *Initially it was the nurse, then it was the physiotherapist, any questions I had for the others would go through her*

F *But, we still had to ask a lot of questions.... initially it was all very disjointed... I think our contact with the Health Board was haphazard until the physiotherapist became our point of contact.*

This theme highlights the value of a multi disciplinary approach to treatment of PWS. Another finding is the significance of a single 'point of contact' in the Early Intervention Services for parents. This provides a clear and effective communication between the family and the team.

Physical effects of PWS – Motor Development

According to the PWSA USA (1999) motor development is delayed typically by one to two years, as are most milestones.⁶ For example, walking usually occurs around age two. Contrary to this, Anne aged three and two months is not walking

⁶ Latson, S.R. (1995) Preventing Parent Burn Out: Model for Teaching Effective Coping Strategies to Parents of Children with Learning Disabilities, Learning Disabilities Association of America. Online.

yet. Anne's father stated that *'she can stand if leaning against something for 20 - 30 seconds'*.

Anne also has a number of pieces of equipment to help her with movement. These are provided by the Early Intervention Services, North Eastern Health Board (now HSE, North East Region).

Physiotherapy is usually the first intervention (from the Early Intervention Services) for children with PWS. This is due to hypotonia (weak muscle tone) being present from early on. A key finding from the theme of is that symptoms and developmental delays can vary greatly for individuals. The study also finds that physiotherapy is an effective form of treatment for motor development and hypotonia.

Hypotonia is a common characteristic in adults with PWS. This condition along with the life-long hyperphagia (insatiable appetite for food) can cause medical complications such as diabetes mellitus and arthritis in the joints. Swimming for children and adults with PWS is a preferable sport which maintains movement of joints without having adverse effects.

Physical effects of PWS – Respiratory complications

According to Loker (2003) *'decreased muscle tone and excessive obesity can decrease the drive for breathing.'*⁷ The most common respiratory problem with PWS is obstructive sleep apnoea – which causes periodic cessations of breathing while the child is asleep.

⁷ Loker, J., M. D., (2003), Respiratory Problems in Prader-Willi Syndrome, in *The Gathered View*, Prader-Willi Syndrome Association (USA).

The first ever sleep studies in relation to sleep apnoea in Ireland commenced in Autumn 2003 in the National Children's Hospital in Tallaght. They commenced in the Spring in the United States. Dr. Meehan explained the purpose of the sleep studies – To see how frequent sleep apnoea occurs. She reported that tonsillectomy procedures are generally carried out if the frequency of apnoeas is high. (A tonsillectomy can help to free up the child's airways, thus reducing the frequency of apnoea).⁸

Anne's parents were asked if Anne suffered from any type of sleep apnoea. F stated – *'Sleep apnoea seems to be linked more with older people, like _____'s son, he's about 20 stone, he suffers from it badly.'* M – *Yes, that's what I thought as well.* As sleep studies had only commenced at time of interview, Anne's parents had not been asked for her to participate at that time. According to Dr. Meehan, 20 children, including some with PWS had participated in the first autumn trial. No results from the sleep studies have been released at the time of writing.

A key finding here is the recent commencement of sleep studies in Ireland, to investigate the cause of sleep apnoea in children with PWS.

Respiratory problems in adults with PWS can cause excessive daytime sleepiness. This sleepiness can be problematic if the person is smoking or carrying out certain work tasks. Supervision by family members or social care workers may be necessary.

Physical effects of PWS – Growth Hormone Therapy

Growth Hormone therapy has been used to treat PWS since the early 1990's. Growth Hormone (GH) is a relatively young therapy; its commercial release was in

⁸ Dr Meehan, J, Paediatrician, National Children's Hospital Tallaght, Phone interview re: Treatment of secondary disorders of PWS and current research, interview held on 3 February 2003.

1985. Nice (2002) reported that *'most children with PWS are obese, the aim of GH therapy is to improve body composition as well as promote growth.'*⁹

Anne's parents commented on her experience of Growth Hormone Therapy (GHT). M - *'Anne has been on growth hormone since May, that's six months now'* (Anne commenced GHT at age 2 years 8 months. Anne grew by 6cm in this time. On average the growth velocity is 6cm to 8cm annually for children on GHT.

Anne's mother mentioned her hopes that Anne would be on GH for the rest of her life. In 2003, NICE for the first time put forward recommendations in relation to GH being administered to adults (to improve hypotonia, bone mass density etc). At the time of writing, Lawlor reports that GH treatment has not yet been sanctioned in Ireland, as a treatment for adults with PWS. She is aware of one Irish male remaining on GH until the age of eighteen, treatment ceased thereafter.¹⁰

A key finding of this theme is that GH deficiency appears to play a role in the short stature characteristic of PWS. GH therapy also has other beneficial effects, on hypotonia and body composition. Another important finding here is the progressive research into treatments for adults with PWS. The effect of GH on adult height has yet to be determined.

Cognitive ability of children with PWS

Most children with PWS are known to have some degree of learning disability. The problem identified in assessing their ability is that characteristics of PWS such as communication delay, speech problems, can mask the level at which the child is operating cognitively.

⁹ The National Institute for Clinical Excellence (NICE) - published guidelines for the use of GH, UK, May 2002.

¹⁰ Lawlor, L - Co founder and Information Officer of Prader-Willi Syndrome Association, Ireland, interviewed held during PWSA Ireland Annual Meeting, on 8 November 2003, Dublin.

It was found in this case study that Anne's strengths lay in concentration and also in long term memory. This is in keeping with findings of other studies (Kleppe et al, 1990 and Hauffa, 2000). Kleppe et al identified word finding as a problem, i.e. the child fully understands what is being said but has difficulty in finding the vocabulary to respond.¹¹

Anne, at the time of writing, has approximately 10 LAMH signs and several verbal sounds, 'mama', 'dada', 'da' for 'dolly' etc. The Early Intervention Services provide LAMH courses for parents of children with speech delay or impairment.¹²

The need for individual programme plans is highlighted in this case study, this being due to the varying strengths and weaknesses of each individual child in relation to learning. Speech therapy is an essential and well-established treatment for children with PWS.

Emotional/Behavioural Effects of PWS

Hypotonia in *infants* with PWS has a clear affect on behaviour. *'The infant with PWS is characteristically passive, quiet, with a weak or absent cry'* (Aughton & Cassidy, in Hauffa 2000: 13).

Anne's mother comments confirmed their statements, M – *'Anne was really floppy...when you look back on it she was totally textbook stuff for Prader Willi.....I think we heard her cry maybe three times in the first year, we'd get really upset, thinking there's something really wrong, we'd set the alarm...for the first 8 weeks we were checking her on the hour...we were paranoid about the feeds as we knew PWS was an eating thing'*

¹¹ Kleppe, C, et al – The Speech and language characteristics of children with PWS, 1990, USA .

¹² Makaton in the UK/LAMH in Ireland is a form of basic sign language, most signs are based on simple actions for e.g. walking two fingers on the palm of the other hand = walking. There are approx 350 signs in LAMH .

The second stage of PWS includes the onset of hyperphagia – insatiable appetite for food. This characteristic has consequent behaviour as explained by Hauffa (2000) *'it is not uncommon for all daily activities to be concentrated on food seeking, hoarding of food is common,...this constant need to eat continues throughout the life of the individual with PWS, and diet can only be controlled by close supervision.'*

Hauffa (2000) cites ritualistic behaviour as a common feature of PWS. This case study found that this trait coupled with the child's interest in food can provide a structure for a strict programme to be put in place in relation to the child's eating habits. Anne's father commented on her eating habits. F- *'I think there could be a lot more flexibility around Anne's diet but we've managed to keep it very structured and we'd hope we can maintain this over the years.'*

This study found that the responsibility lies wholly on the parents of children with PWS to be pro-active in planning and implementing routine dietary programmes thus setting the stage for life long habits.

The insatiable appetite for food and ritualistic behaviour in childhood and adulthood can be a challenge for family and carers. A strict programme in relation to access to food is needed but care is needed not to exclude the person from family or group home meals.

Effects of PWS on Social Skills of the Child

Children with PWS are described as being good natured, placid and co-operative (PWUSA UK, 1999). They are also described as having severe temper tantrums, being aggressive in relation to wanting access to food. Some of the common traits among children with PWS can impinge on their social skills.

The fact that children with PWS are quite physically restricted in their movement can lead to the child being isolated from their peers. Anne's father commented – *'Anne can't wander over to anybody like the other kids (in pre-school setting), she does her own thing a lot, she'd stay playing with toys for 20 – 30 minutes.'* This highlights how Anne's concentration is a key skill in her development and quality of life.

Hyperphagia, associated with PWS is said to set in between the ages of 1 – 4 years old. This can be difficult at pre-school age; other children's lunches can become a target. This study found that the parent's role as an educator is fundamental in ensuring other people in the child's life, extended family, teachers, caregivers etc, adhere to strict dietary control, and not give in to temptation, i.e. giving the child extra food, providing treats etc.

The PWSA UK (1999) that *Parents too, may feel isolated, because there are so few families in any one area who have a child who has PWS. The unsociable aspect of the eating disorder can make parents feel isolated from the rest of society.*

In this study Anne's father talked about the support they receive from Anne's mother's sisters who live close by. He stated that *'we (husband and wife) always try to look at the positive things and just enjoy Anne while she is young as the children grow up so fast.'*

Recommendations

1. Due to this case study finding that non-specialist medical staff may not have previous knowledge of PWS prior to a presenting case, it is recommended that a Genetic Counsellor be available to parents at time of diagnosis, preferably in the birth hospital.
2. Following discussion of *practice* with Respiratory Paediatrician Tallaght Hospital in relation to a child starting Growth Hormone Treatment and the findings of Janalee Heinemann (2003), it is recommended that a child

with PWS should have a sleep study test and also an Ear, Nose and Throat (ENT) examination prior to commencement of Growth Hormone Treatment (GHT).

Endnotes

This research paper was submitted as part of the BA (Hons) in Social Care at the Cork Institute of Technology April 2004.

Note regarding physical effects of PWS – Respiratory complications

Prior to the publication of this paper, results of sleep studies on children with PWS, (carried out in Temple Street Hospital) were published in November 2004. The study entitled *Sleep Abnormalities in children with Prader Willi Syndrome* found that 82% of the children with PWS had abnormalities which consisted of central or obstructive apnoeas to mixed apnoeas. Intervention was needed in all children with abnormalities. These interventions included tonsillectomy and nasal ventilation. The study therefore recommended that sleep studies should be carried out on all children with PWS as part of their management routine (Irish Journal of Medical Science, 2004).

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